



HGSNAT gene

heparan-alpha-glucosaminide N-acetyltransferase

Normal Function

The *HGSNAT* gene provides instructions for producing an enzyme called heparan-alpha-glucosaminide N-acetyltransferase (which is often shortened to N-acetyltransferase). This enzyme is located in lysosomes, compartments within cells that digest and recycle different types of molecules. N-acetyltransferase is involved in the step-wise breakdown (degradation) of large molecules called glycosaminoglycans (GAGs). GAGs are composed of sugar molecules that are linked together to form a long string. To break down these large molecules, individual sugars are removed one at a time from one end of the molecule. N-acetyltransferase adds a molecule called an acetyl group to the sugar glucosamine in a subset of GAGs called heparan sulfate. This addition prepares the GAG for the next step in the degradation process.

Health Conditions Related to Genetic Changes

mucopolysaccharidosis type III

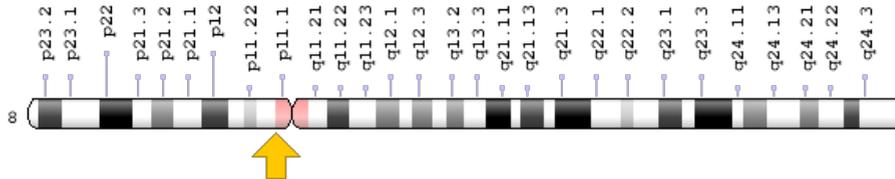
At least 54 mutations in the *HGSNAT* gene have been found to cause mucopolysaccharidosis type IIIC (MPS IIIC). Most of these mutations change single DNA building blocks (nucleotides) in the gene. All of the mutations that cause MPS IIIC reduce or eliminate the function of N-acetyltransferase.

The lack of N-acetyltransferase activity disrupts the breakdown of heparan sulfate. As a result, partially broken down heparan sulfate accumulates within lysosomes. Researchers believe that the accumulation of GAGs interferes with the functions of other proteins inside the lysosomes and disrupts the normal functions of cells. It is unknown why the buildup of heparan sulfate mostly affects the central nervous system in MPS IIIC.

Chromosomal Location

Cytogenetic Location: 8p11.21-p11.1, which is the short (p) arm of chromosome 8 between positions 11.21 and 11.1

Molecular Location: base pairs 43,140,449 to 43,202,827 on chromosome 8 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- acetyl coenzyme A:alpha-glucosaminide N-acetyltransferase
- DKFZp686G24175
- FLJ22242
- FLJ32731
- HGNAT
- HGNAT_HUMAN
- TMEM76
- transmembrane protein 76

Additional Information & Resources

Educational Resources

- Eureka Bioscience Collection: Defects in Glycosaminoglycan Degradation (Mucopolysaccharidoses)
<https://www.ncbi.nlm.nih.gov/books/NBK6177/#A53462>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28HGSNAT%5BTIAB%5D%29+OR+%28%28alpha-glucosaminide+N-acetyltransferase%5BTIAB%5D%29+OR+%28HGNAT%5BTIAB%5D%29+OR+%28TMEM76%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- HEPARAN-ALPHA-GLUCOSAMINIDE N-ACETYLTRANSFERASE
<http://omim.org/entry/610453>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_HGSNAT.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=HGSNAT%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=26527
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/138050>
- UniProt
<http://www.uniprot.org/uniprot/Q68CP4>

Sources for This Summary

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Reviewed: August 2010

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
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